

SINGLE GENE DISORDERS

Test Required	Specimen Type	Container	Volume	Transport/Storage
DNA Test, Single gene, Gene panel or Exome	Blood	EDTA	3ml Adults/children 1ml newborn	1 st Class post or courier

POSTNATAL TESTING

Test Required	Specimen Type	Container	Volume	Transport/Storage
Karyotype and/or FISH	Blood	Lithium Heparin	2ml Adults/children 1ml newborn	1 st Class post or courier
Array/QF-PCR	Blood	EDTA	3ml Adults/children 1ml newborn	1 st Class post or courier (taxi for urgents)

PRENATAL TESTING

Test Required	Specimen Type	Container	Volume	Transport/Storage
Rapid QF-PCR, Array, Karyotype, Single gene (Parental bloods in lith hep and EDTA should be sent with prenatal samples)	Amnio	Sterile Universal	2ml Adults/children 1ml newborn	Courier or Taxi. Must arrive by 14.00 for same day processing (refrigerate overnight if cannot have arrive before laboratory closing)
	CVS	Sterile Universal with transport media (available from laboratory)	3ml Adults/children 1ml newborn	

TISSUES

Test Required	Specimen Type	Container	Volume	Transport/Storage
QF-PCR, Array, DNA storage	POC, placenta/cord, fetal tissue biopsy	Sterile Universal with saline/transport media		SEND TO PATHOLOGY NRIE
Mosaicism score/Cryopreservation	Skin biopsy/other solid tissues			SEND TO GLASGOW (West of Scotland Genetics)

ONCOLOGY

Test Required	Specimen Type	Container	Volume	Transport/Storage
Karyotype, FISH, Array	Bone Marrow Aspirate (Blood for CLL FISH etc)	Sterile Universal with BM/biopsy media, available from laboratory (or Lithium Heparin blood tube)	Attempted processing of all sizes of samples.	Courier or Taxi (Phone to inform laboratory before sending urgents)
Chromosome Breakage	Blood	Lithium Heparin and EDTA		SEND TO ABERDEEN (North of Scotland Genetics)



Lithium heparin
(sometimes labelled
LH)



EDTA
(sometimes labelled
K2)